



NPC2 gene

NPC intracellular cholesterol transporter 2

Normal Function

The *NPC2* gene provides instructions for making a protein that is located inside lysosomes, which are compartments in the cell that digest and recycle materials. The NPC2 protein attaches (binds) to cholesterol. Cholesterol is a waxy, fat-like substance that is produced in the body and obtained from foods. Research suggests that the NPC2 protein plays an important role in moving cholesterol and certain other fats (lipids) out of the lysosomes to other parts of the cell.

Health Conditions Related to Genetic Changes

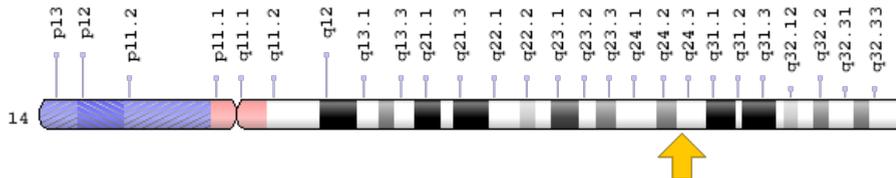
Niemann-Pick disease

More than 20 mutations in the *NPC2* gene have been found to cause Niemann-Pick disease type C2. This type of Niemann-Pick disease is characterized by a buildup of fat within cells that leads to movement problems, neurological impairment, lung and liver disease, and speech and feeding problems. The *NPC2* gene mutations that cause Niemann-Pick disease type C2 reduce or eliminate NPC2 protein activity, which prevents movement of cholesterol and other lipids, leading to their accumulation in cells. Because these lipids are not in their proper location in cells, many normal cell functions that require lipids (such as cell membrane formation) are impaired. The accumulation of lipids and the cell dysfunction eventually leads to cell death, causing the tissue and organ damage seen in Niemann-Pick disease type C2.

Chromosomal Location

Cytogenetic Location: 14q24.3, which is the long (q) arm of chromosome 14 at position 24.3

Molecular Location: base pairs 74,479,940 to 74,493,475 on chromosome 14 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- epididymal secretory protein
- HE1
- MGC1333
- Niemann-Pick disease, type C2
- NP-C2
- NPC2_HUMAN

Additional Information & Resources

Educational Resources

- Madame Curie Bioscience Database: Defects in Lipid Degradation
<https://www.ncbi.nlm.nih.gov/books/NBK6177/#A53465>

GeneReviews

- Niemann-Pick Disease Type C
<https://www.ncbi.nlm.nih.gov/books/NBK1296>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NPC2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- NPC2 GENE
<http://omim.org/entry/601015>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_NPC2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=NPC2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=14537
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/10577>
- UniProt
<http://www.uniprot.org/uniprot/P61916>

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